



## ADNP gene

activity dependent neuroprotector homeobox

### Normal Function

The *ADNP* gene provides instructions for making a protein that helps control the activity (expression) of other genes through a process called chromatin remodeling. Chromatin is the network of DNA and protein that packages DNA into chromosomes. The structure of chromatin can be changed (remodeled) to alter how tightly DNA is packaged. Chromatin remodeling is one way gene expression is regulated during development; when DNA is tightly packed, gene expression is lower than when DNA is loosely packed. As part of the remodeling process, the ADNP protein attaches to DNA and interacts with groups of proteins called SWI/SNF complexes, which direct changes in the structure of chromatin.

By regulating gene expression, the ADNP protein is involved in many aspects of development. It is particularly important for regulation of genes involved in normal brain development, and it likely controls the activity of genes that direct the development and function of other body systems.

### Health Conditions Related to Genetic Changes

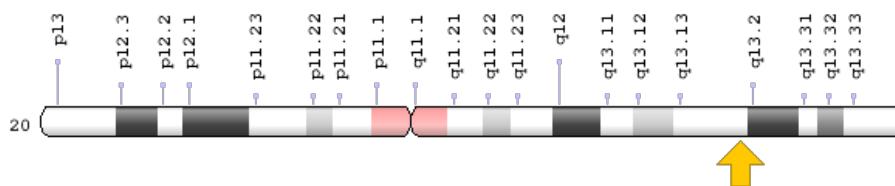
#### ADNP syndrome

At least 22 *ADNP* gene mutations have been found to cause *ADNP* syndrome. This condition features intellectual disability and autism spectrum disorder, which is characterized by impaired communication and social interactions. Affected individuals can also have distinctive facial features and a wide variety of other signs and symptoms. Most *ADNP* gene mutations are thought to lead to the production of an abnormally short ADNP protein. Although it is unclear how these genetic changes cause *ADNP* syndrome, researchers speculate that the abnormally short protein can attach to DNA but cannot interact with SWI/SNF complexes. As a result, chromatin remodeling is impaired. Disturbance of this process alters the activity of many genes and disrupts the development or function of several of the body's tissues and organs, including the brain. These changes likely explain the intellectual disability, autism spectrum disorder, and other diverse signs and symptoms of *ADNP* syndrome.

## Chromosomal Location

Cytogenetic Location: 20q13.13, which is the long (q) arm of chromosome 20 at position 13.13

Molecular Location: base pairs 50,888,918 to 50,934,984 on chromosome 20 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- activity-dependent neuroprotective protein
- activity-dependent neuroprotector
- activity-dependent neuroprotector homeobox protein
- ADNP homeobox 1
- ADNP1
- HVDAS
- KIAA0784
- MRD28

## Additional Information & Resources

### Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): ATP-driven Chromatin Remodeling Machines Change Nucleosome Structure  
<https://www.ncbi.nlm.nih.gov/books/NBK26834/#A644>
- Molecular Biology of the Cell (fourth edition, 2002): Chromosomal DNA and Its Packaging in the Chromatin Fiber  
<https://www.ncbi.nlm.nih.gov/books/NBK26834/>

## Scientific Articles on PubMed

- PubMed

<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28ADNP%5BTIAB%5D%29+OR+%28activity+dependent+neuroprotector+homeobox%5BTIAB%5D%29%29+OR+%28%28ADNP+homeobox+1%5BTIAB%5D%29+OR+%28activity-dependent+neuroprotective+protein%5BTIAB%5D%29+OR+%28activity-dependent+neuroprotector+homeobox+protein%5BTIAB%5D%29+OR+%28activity-dependent+neuroprotector%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

## OMIM

- ACTIVITY-DEPENDENT NEUROPROTECTOR HOMEobox  
<http://omim.org/entry/611386>

## Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_ADNP.html](http://atlasgeneticsoncology.org/Genes/GC_ADNP.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=ADNP%5Bgene%5D>
- HGNC Gene Family: ZF class homeoboxes and pseudogenes  
<http://www.genenames.org/cgi-bin/genefamilies/set/529>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=15766](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=15766)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/23394>
- UniProt  
<http://www.uniprot.org/uniprot/Q9H2P0>

## **Sources for This Summary**

- OMIM: ACTIVITY-DEPENDENT NEUROPROTECTOR HOMEobox  
<http://omim.org/entry/611386>
- Helsmoortel C, Vulto-van Silfhout AT, Coe BP, Vandeweyer G, Rooms L, van den Ende J, Schuurs-Hoeijmakers JH, Marcelis CL, Willemsen MH, Vissers LE, Yntema HG, Bakshi M, Wilson M, Witherspoon KT, Malmgren H, Nordgren A, Annerén G, Fichera M, Bosco P, Romano C, de Vries BB, Kleefstra T, Kooy RF, Eichler EE, Van der Aa N. A SWI/SNF-related autism syndrome caused by de novo mutations in ADNP. *Nat Genet.* 2014 Apr;46(4):380-4. doi: 10.1038/ng.2899.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/24531329>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3990853/>

- Malishkevich A, Leyk J, Goldbaum O, Richter-Landsberg C, Gozes I. ADNP/ADNP2 expression in oligodendrocytes: implication for myelin-related neurodevelopment. *J Mol Neurosci.* 2015 Oct;57(2):304-13. doi: 10.1007/s12031-015-0640-4.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/26315608>
- Mandel S, Rechavi G, Gozes I. Activity-dependent neuroprotective protein (ADNP) differentially interacts with chromatin to regulate genes essential for embryogenesis. *Dev Biol.* 2007 Mar 15; 303(2):814-24.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/17222401>
- Mandel S, Spivak-Pohis I, Gozes I. ADNP differential nucleus/cytoplasm localization in neurons suggests multiple roles in neuronal differentiation and maintenance. *J Mol Neurosci.* 2008 Jun; 35(2):127-41. doi: 10.1007/s12031-007-9013-y.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/18286385>
- Vandeweyer G, Helsmoortel C, Van Dijck A, Vulto-van Silfhout AT, Coe BP, Bernier R, Gerdts J, Rooms L, van den Ende J, Bakshi M, Wilson M, Nordgren A, Hendon LG, Abdulrahman OA, Romano C, de Vries BB, Kleefstra T, Eichler EE, Van der Aa N, Kooy RF. The transcriptional regulator ADNP links the BAF (SWI/SNF) complexes with autism. *Am J Med Genet C Semin Med Genet.* 2014 Sep;166C(3):315-26. doi: 10.1002/ajmg.c.31413.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/25169753>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4195434/>

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